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Twenty-year clinical progression of dysferlinopathy in patients from Dagestan

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Abstract

© 2017 Umakhanova, Bardakov, Mavlikeev, Chernova, Magomedova, Akhmedova, Yakovlev, Dalgatov, Fedotov, Isaev and Deev. To date, over 30 genes with mutations causing limb-girdle muscle dystrophy have been described. Dysferlinopathies are a form of limb-girdle muscle dystrophy type 2B with an incidence ranging from 1:1,300 to 1:200,000 in different populations. In 1996, Dr. S. N. Illarioshkin described a family from the Botlikhsky district of Dagestan, where limb-girdle muscle dystrophy type 2B and Miyoshi myopathy were diagnosed in 12 members from three generations of a large Avar family. In 2000, a previously undescribed mutation in the DYSF gene (c.TG573/574AT; p. Val67Asp) was detected in the affected members of this family. Twenty years later, in this work, we re-examine five known and seven newly affected family members previously diagnosed with dysferlinopathy. We observed disease progression in family members who were previously diagnosed and noted obvious clinical polymorphism of the disease. A typical clinical case is provided.

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Keywords

Dysferlin, Dysferlinopathy, LGMD2B, Miyoshi myopathy, Muscular dystrophy

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